CYP27B1 gene

cytochrome P450 family 27 subfamily B member 1

Normal Function

The *CYP27B1* gene provides instructions for making an enzyme called 1-alpha-hydroxylase (1α-hydroxylase). This enzyme carries out the final reaction to convert vitamin D to its active form, 1,25-dihydroxyvitamin D_3, also known as calcitriol. Vitamin D can be acquired from foods in the diet or can be made in the body with the help of sunlight. When active, this vitamin is involved in maintaining the proper balance of several minerals in the body, including calcium and phosphate, which are essential for the normal formation of bones and teeth. One of vitamin D's major roles is to control the absorption of calcium and phosphate from the intestines into the bloodstream. Vitamin D is also involved in several process unrelated to bone formation.

In order to carry out the roles of vitamin D, calcitriol attaches (binds) to another protein known as vitamin D receptor (VDR). The resulting calcitriol-VDR complex then binds to particular regions of DNA, known as vitamin D response elements, and regulates the activity of vitamin D-responsive genes. By turning the genes on or off, VDR helps control calcium and phosphate absorption and other processes.

Health Conditions Related to Genetic Changes

autoimmune Addison disease

multiple sclerosis

vitamin D-dependent rickets

Mutations in the *CYP27B1* gene cause vitamin D-dependent rickets type 1 (VDDR1), also known as vitamin D 1α-hydroxylase deficiency. This disorder of bone development is characterized by low levels of calcium (hypocalcemia) and phosphate (hypophosphatemia) in the blood, which lead to soft, weak bones that are prone to fracture. A common feature of this condition is bowed legs.

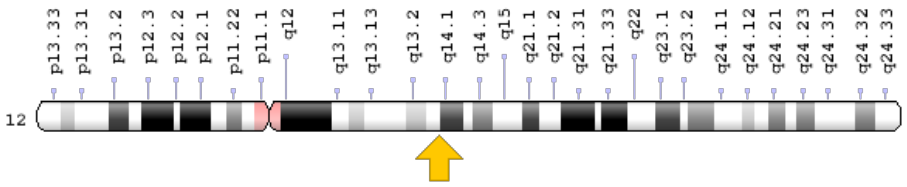
The *CYP27B1* gene mutations that cause this condition reduce or eliminate the function of 1α-hydroxylase. As a result, vitamin D does not get converted to its active form and cannot stimulate the activity of genes important for mineral absorption. The lack of calcium and phosphate absorption from the intestines into the blood slows the deposition of these minerals in developing bones (bone mineralization), which leads

to soft, weak bones and other features of VDDR1. Hypocalcemia also causes muscle weakness and seizures in some affected individuals.

Chromosomal Location

Cytogenetic Location: 12q14.1, which is the long (q) arm of chromosome 12 at position 14.1

Molecular Location: base pairs 57,762,334 to 57,767,193 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 1alpha(OH)ase
- 25-hydroxyvitamin D-1 alpha hydroxylase, mitochondrial
- 25 hydroxyvitamin D3-1-alpha hydroxylase
- 25-OHD-1 alpha-hydroxylase
- CYP1alpha
- CYP27B
- cytochrome p450 27B1
- cytochrome P450 subfamily XXVIIB polypeptide 1
- cytochrome P450, family 27, subfamily B, polypeptide 1
- cytochrome P450C1 alpha
- cytochrome P450VD1-alpha
- P450c1
- VD3 1A hydroxylase

Additional Information & Resources

Educational Resources

- Dietary Reference Intakes for Calcium, Phosphorus, Magnesium, Vitamin D, and Fluoride (1997): Vitamin D
  https://www.ncbi.nlm.nih.gov/books/NBK109831/
- Endocrinology: An Integrated Approach (2001): Classical Actions of Vitamin D on Intestine and Bone
  https://www.ncbi.nlm.nih.gov/books/NBK24/#_A788_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CYP27B1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- CYTOCHROME P450, SUBFAMILY XXVIIB, POLYPEPTIDE 1
  http://omim.org/entry/609506

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CYP27B1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CYP27B1%5Bgene%5D
- HGNC Gene Family: Cytochrome P450 family 27
  http://www.genenames.org/cgi-bin/genefamilies/set/1014
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2606
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1594
- UniProt
  http://www.uniprot.org/uniprot/O15528

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